

Pompe Disease

What are the Symptoms of Pompe Disease?

Muscle weakness is one of the main symptoms for Pompe disease and is evident in both the infantile- and late-onset forms of the disorder. In addition to mobility, muscle weakness will affect the respiratory system to the degree that mechanical ventilation may become necessary. Some people find they have morning headaches and have trouble concentrating due to the lack of oxygen. Other symptoms associated with late-onset Pompe disease include difficulty with chewing and swallowing; lower back pain; scoliosis; and frequent falls.

What is Pompe Disease?

Pompe Disease occurs in many different age groups, anywhere from babies to adults, and is classified as a rare neuromuscular disorder. While the most common name may be Pompe disease, after Dr. J.C. Pompe, the first man to have described this disorder back in 1932, it can also go by Acid Maltase Deficiency and Glycogen Storage Disease type 2.

If a child is diagnosed with Pompe disease within the first year of life, it is generally considered to be the infantile-onset form of the disease and in most cases the disease is so progressive it becomes fatal before their first birthday. If a child is diagnosed after the first year of life they may get the diagnosis of having late-onset Pompe disease that, while still progressive, progresses much slower than infantile-onset and can also be diagnosed during adult hood. Approximately one third of people diagnosed with Pompe disease are diagnosed with infantile-onset while the other two thirds are diagnosed with late-onset.

In infantile-onset Pompe disease, the infant is too weak to breathe on its own or to suckle its milk. The heart becomes enlarged, growing to three times its normal size. This reduces the size of the heart's chambers, forcing it to pump much harder. Other symptoms of infantile-onset Pompe disease include the failure to gain weight; enlarged tongue and liver; frog-like position of legs; and floppiness due to lack of muscle tone.

How Does Pompe Disease Progress?

Pompe disease is a progressive disorder. As with most neuromuscular diseases, Pompe disease begins with a loss of muscle control. For infants there is a lack of the basic motor skills such as learning to hold their head up, walking and crawling. For adults they begin with having difficulty walking and, eventually, difficulty climbing stairs.

Respiratory symptoms begin with slight difficulty breathing leading in many cases, as described above, to mechanical ventilation.

Pompe disease progresses at different rates among those affected by the disorder.

How is Pompe Disease Diagnosed?

Pompe disease can be hard to diagnose as other neuromuscular disorders share many of the same symptoms. For example, Duchenne muscular dystrophy shares progressive muscle weakness, respiratory impairment and difficulty walking with Pompe disease. To confirm a diagnosis of Pompe disease a test that will measure acid alpha-glucosidase enzyme activity can be ordered. This can be done through blood samples or more invasive procedures such as a skin or a muscle biopsy.

Individuals whose tests come back with a relatively high level of acid alpha-glucosidase will need to get them analyzed to determine if they are a carrier of the defective gene or actually have Pompe disease.

How Disabling is Pompe Disease?

Pompe disease usually has a negative effect on life spans, especially in the case of infant-onset. For late-onset though there are many aides to help make everyday life easier such as wheelchairs and ventilation machines when necessary. While a person's lifespan might be shortened, they can still live a fairly typical life.

What is the Age of Onset?

Age of onset varies greatly. In the early Infant-onset kind of Pompe, it can happen between a few weeks to a few months from birth. For late-onset, symptoms may be delayed until the person is in their 40's or 50's.

Research

There is much research focused on infantile-onset. Great promise is appearing in research and tests being done using enzyme replacement therapy. This involves injecting acid alpha-glucosidase directly into the blood streams of affected infants. In Europe the use of this gene therapy has been approved for patients regardless of age.

Symptom Management

There are several different ways to manage the symptoms of Pompe disease. Starting with a specific diet plan can help. Seeing a dietician will provide knowledge on what kinds of foods are best. Physical therapy will help keep muscles mobile and will help manage other symptoms of Pompe disease. Occupational therapy can help people learn use different adaptive devices in an effort to make everyday living easier. When breathing becomes difficult, the use of a ventilation machine can be prescribed.

What causes Pompe Disease?

Pompe disease is caused through a genetic defect in the chromosomes. Everybody has 23 pairs of chromosomes. Pompe disease is caused when there is a defect on both chromosomes of the 17th set. Since the child gets one chromosome from each parent, both parents have to have the defective chromosome to give their child. If only one of the two chromosomes from the 17th spot is defective in the child, then it will simply become a carrier like its parents were. There is also the possibility that the child will not inherit any defective gene from their parents and in this case they have no chance of giving their future children the gene. Basically, if there are two carrier parents there is a 25% chance that the child will have Pompe disease.

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